Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: September 9, 2014, 2:00 p.m.

Subcommittee Members Attending:

Lurie Children's Hospital	Barbara Burton, Chair
	Joel Charrow
St. Louis Children's Hospital	Kathy Grange
University of Chicago	Darrel Waggoner
University of Illinois at Chicago	George Hoganson
	Annie McRae
University of Illinois Chicago at Peoria	Jennifer Burton
DSCC	Tess Rhodes

IDPH Staff:

Khaja Basheeruddin	Tom Johnson
Jean Becker	Arthur Kohrman
Marie Bukovic	Claudia Nash
Matt Charles	Elizabeth Paton
Maria Crain	Tom Schafer
George Dizikes	Rong Shao
Shannon Harrison	Heather Shryock

A special call convened to discuss the delays of the LSD pilot and Krabbe testing.

IDPH Report

Dr. Dizikes explained the original intent was for molecular testing of Krabbe to be done by the New York State Health Department Wadsworth Center, but they have rejected the contract due to indemnification language. Elizabeth Paton, IDPH Legal Counsel, is working with the state's Department of Central Management Services (CMS) who oversees contractual agreements within the state. Due to the financial structure used to reimburse New York State Department of Health Wadsworth Center, it is likely that indemnification language required by New York, but rejected by Illinois CMS attorney will prevent finalization of a contract for providing Krabbe mutation analysis from dried blood spots. In the interim, Illinois does have the ability to enter into a two party agreement up to $50,000 with Mayo Clinic to do the molecular testing, but the rate per test is nearly twice that of New York. By delaying the start of Krabbe screening until Jan. 1, 2015, this would allow IDPH to get a two-party agreement in place with Mayo Clinic and to prepare a bid to procure Krabbe DNA testing. If the bid process has not been completed by the time the $50,000 with Mayo Clinic has been expended, there are options that would permit Krabbe molecular testing to continue without interruption.

Furthermore, Dr. George Dizikes discussed various equipment issues. The high pressure pumps on the mass spectrometers are malfunctioning due to leaky seals. Currently, the lab is not able to have more than 2-3 machines operational for more than a few days. The lab is getting a fifth

mass spectrometer and hopes to have four machines operational at any one time to be able to accommodate going statewide.

Additional discussion ensued around incorporating psychosine testing for Krabbe, but it was decided that the protocol was developed to include the molecular component, and protocols would need to be revised to consider triaging cases differently. There is no current published literature to support psychosine as an indicator and predictor of development of clinical symptoms. Dr. Dizikes indicated that if enzyme testing was the only screening test done, it is expected to result in 100 abnormals per year creating a significant burden on specialists who would be required to see 1-2 of these patients a week.

Dr. Hoganson referred to the legislation for newborn screening which dictates an "appropriate test" must be in place to do screening. Dr. Waggoner also voiced that the Laboratory Subcommittee must act responsibly and questioned if it was reasonable to screen based solely on enzyme levels.

Based on the issues at hand, Deputy Director Tom Schafer recommended delaying the start of Krabbe testing until January 2015 while the Department prepares a bid to procure Krabbe DNA testing from an outside laboratory.

Other concerns involved the database vendor, Perkin Elmer. They are working to make corrections to the database necessary for the Follow-Up Program to be able to report abnormal results appropriately.

The meeting adjourned 3:00 p.m.

Next scheduled call is October 16, 2014 at 2:00 PM
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